MUTATIONS IN HUMAN GLUTAMATE CARBOXYPEPTIDASE II GENE IMPACTING FOLATE METABOLISM, AND DETECTION OF AFFECTED INDIVIDUALS

ABSTRACT OF THE DISCLOSURE

The invention provides methods for detecting mutations in the human GCPII gene which affect the ability of an individual to hydrolyze terminal glutamates from dietary folates. Such individuals are at increased risk for conditions associated with hyperhomocyteinemia, in particular, cardiovascular disease, colon cancer, and altered cognition in the elderly, including Alzheimer's disease. In addition, pregnant women with low folate status are at increased risk of bearing children with neural tube defects and congenital heart defects. Individuals with these mutations can be screened and treated with supplementation of their diet with folic acid.